Undescended testis with congenital unilateral absence of the vas deferens

V.D. Kulkarni, N.A. Deshpande, P.N. Deshpande, M. Phanse
Department of Surgery, Y.C.M. Hospital, Pimpri Pune, India.

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Abstract

Congenital absence of the vas deferens (CAVD) is a condition in which the vasa deferentia fail to form properly prior to birth. It may either be unilateral (CUAVD) or bilateral (CBAVD). Congenital absence of vas deferens is responsible for up to 2% of male infertility. The disease is considered to be a primary genital form of cystic fibrosis. It contributes towards 6% of obstructive azoospermia cases. Up to 80% of patients with CAVD have mutations associated with Cystic Fibrosis Transmembrane Conductance (CFTR) gene. Upper urinary tract malformations are noted in up to 20% of cases. This category of patients have a normal frequency of CFTR mutations and considered a distinct clinical entity. We hereby report a rare case of CUAVD with ipsilateral renal agenesis with cryptorchidism.

Introduction

Disturbances in the development of human embryo are responsible for the absence of vas deferens. Vas deferens is derived from the mesonephric duct while the testis and the globus major of the epididymis arise from the genital ridge. The extent of abnormalities depends on the time at which mesonephric duct stops developing. If it occurs early in embryonic life (around the fourth week) the ureter and kidney are absent [1]. If the disturbances happen beyond fourth week, the kidney and ureter are present, although their development could be abnormal [2].

Case History

A 12 year old child presented with non-descent of the right testis since birth. He did not have associated abdominal pain. On examination, the right testis was neither palpable in the scrotum nor found in the right inguinal region. The left testis was normal in size and the cord was palpable on scrotal examination. There was no report of hospital admissions in the past. Ultrasonography of the abdomen revealed an absent right kidney, the left kidney being normal in size and shape. The right testis was detected, by ultrasound, in the right inguinal region. Intravenous pyelography confirmed the above findings (Figure 1). Renal function tests were normal. Sweat sodium estimation was in the normal range. The patient received an orchidopexy after informed consent was obtained from his parents. Intraoperatively the right testis was in inguinal canal (Figure 2). On evaluating cord structures, the vas deferens was absent. Following orchidopexy, he was discharged on the second day.

Discussion

Abnormalities in the vas deferens depend on the time at which the mesonephric duct stops developing. Renal agenesis associated with CUAVD occurs if the
mesonephric duct stops developing by the fourth week [1]. The vas deferens is a cord like structure which can be palpated easily in the normal scrotum. CA VD is a distinct clinical entity. The diagnosis is easily made on scrotal examination [3]. A lower frequency of CFTR mutations has been detected in patients with CUA VD [4]. Between 11% and 26% of patients with CA VD have renal agenesis [5]. The association of CA VD with renal agenesis in cryptorchidism is very low. CFTR mutations are detected in men with CBA VD and, with a lesser frequency in men with CA VD, it appears that a normal amount of functional CFTR protein is required to ensure proper development of the vas deferens. However the lack of CFTR mutations detected in CA VD with renal anomalies also suggest that the mesonephric duct plays a role in the development of the vas deferens. CA VD is the cause of male infertility and hence genetic counseling plays a major role in patients with CA VD [5].

Conclusions
CUAVD associated with renal anomalies and cryptorchidism is a rare diagnosis. CUAVD may be associated with cystic fibrosis and evaluation of the same needs to be considered in all such patients. CAVD can cause infertility in men. Genetic counseling plays a major role in patients with CAVD because of its intricate relations with CFTR gene.

References

Key points:
- CUAVD can be associated with renal anomalies and cryptorchidism is a rare diagnosis.
- CA VD can be a cause of infertility in males.